



## I. BACKGROUND

Amelia Stone received a DTaP<sup>1</sup> vaccination on August 27, 2001, shortly after her birth on April 17, 2001. (*See* Decision of Special Master Golkiewicz at 4, Apr. 15, 2010 (“Dec.”).) Shortly after receiving the vaccination, Amelia experienced two severe seizure episodes. (*See id.* at 4-5.) Amelia was subsequently diagnosed with SMEI, a “particular epilepsy syndrome” that “begins in the first year of life in previously healthy children” and may result in “long lasting [seizures] . . . associated with fever.” (*Id.* at 2 n.2.) “SMEI” stands for “Severe Myoclonic Epilepsy of Infancy,” also known as “Dravet Syndrome.” (*Id.*) As a result of her SMEI, Amelia continued to experience seizures, which have led to developmental delay. (*See id.* at 5-6.)

Amelia’s parents filed a petition for compensation on Amelia’s behalf under the Vaccine Act. At trial, respondent, the Secretary of the Department of Health and Human Services, relied on testimony from Dr. Gerald Raymond, a pediatric neurologist and geneticist. (*See id.* at 13.) Dr. Raymond testified that: “Amelia’s vaccinations neither caused nor exacerbated her SMEI, but rather a mutation in her SCN1A gene is solely responsible for her SMEI.” (*Id.* at 18.) Petitioners relied on testimony from Dr. Marcel Kinsbourne, a neurologist. (*See id.* at 11.) Dr. Kinsbourne acknowledged that “the SCN1A gene mutation plays a role in Amelia’s SMEI,” but claimed that the vaccine “triggered the onset of her Dravet syndrome” by “lower[ing] her seizure threshold.” (*Id.* at 40.) After hearing the parties’ testimony and arguments, the special master found in favor of respondent and entered an order denying compensation. (*See id.* at 51-52.)

This Court subsequently reversed the order on October 28, 2010, holding that the special master failed to apply the correct legal standard for causation. (*See* Order on Mot. for Review.) On remand, the special master once again denied compensation, finding that respondent had “prove[n] by a preponderance of the evidence that the SCN1A gene mutation was the sole cause and that it was principally responsible for Amelia’s SMEI.”<sup>2</sup> (Dec. on Remand at 2.)

## II. STANDARD OF REVIEW

The Court “owe[s] no deference to the . . . special master on questions of law;” such questions are subject to de novo review. *Broekelschen v. Sec’y of Health & Human Servs.*, 618 F.3d 1339, 1345 (Fed. Cir. 2010). However, the Court must “uphold the special master’s findings of fact unless they are arbitrary and capricious.” *Id.* The Court does not “reweigh the factual evidence, or [ ] assess whether the special master correctly evaluated the evidence,” nor does the Court “examine the probative value of the evidence or the credibility of the witnesses.” *Id.* at 1349 (quoting *Munn v. Sec’y of Health & Human Servs.*, 970 F.2d 863, 871 (Fed. Cir.

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<sup>1</sup> “DTaP” stands for “Diphtheria-Tetanus-acellular-Pertussis.” (Dec. at 2.)

<sup>2</sup> The special master’s decision on remand incorporates by reference the findings and discussions from his previous decision, to the extent those findings and discussions are consistent with the Court’s remand instructions. (*See* Dec. on Remand at 2.) Accordingly, the Court cites to the special master’s previous decision where appropriate.

1992)). “[If] the Special Master’s conclusion [is] based on evidence in the record that [is] not wholly implausible, [the Court is] compelled to uphold that finding as not being arbitrary or capricious.” *Cedillo v. Sec’y of Health & Human Servs.*, 617 F.3d 1328, 1338 (Fed. Cir. 2010) (quoting *Lampe v. Sec’y of Health & Human Servs.*, 219 F.3d 1357, 1363 (Fed. Cir. 2000)). Moreover, “[t]he special master’s decision often times is based on the credibility of the experts and the relative persuasiveness of their competing theories;” the special master’s findings with respect to expert credibility “are virtually unchallengeable on appeal.” *Broekelschen*, 618 F.3d at 1347 (quoting *Lampe*, 219 F.3d at 1362).

### III. ANALYSIS

#### A. SCN1A Mutation as the Sole Substantial Cause of Amelia’s SMEI

Petitioners argue that relevant medical literature and Dr. Raymond’s admissions show that the knowledge necessary to make a prediction about the effects of Amelia’s SCN1A mutation does not exist. Respondent argues that it presented reliable, preponderant evidence which established that a factor unrelated was responsible for Amelia’s SMEI.<sup>3</sup>

As a threshold matter, petitioners take issue with Dr. Raymond’s qualifications, arguing that he is “**not** an SCN1A expert,” “[n]or is he an expert in epilepsy.” (Mot. at 29 (emphasis in original).) Dr. Raymond is, however, “an associate professor of neurology at Johns Hopkins University and the director of neurogenetics at the Kennedy Krieger Institute, an affiliate of Johns Hopkins medical School.” (Dec. at 13.) Further, Dr. Raymond “performs consulting services in genetics at ... Johns Hopkins,” and “provides genetics counseling and consultation to families in relation to the results of genetics tests.” (*Id.*) Based on these credentials, the special master was not arbitrary or capricious in determining that Dr. Raymond was qualified to testify on the relationship between Amelia’s SCN1A mutation and her SMEI. (*Id.*) See *SEB S.A. v. Montgomery Ward & Co., Inc.*, 594 F.3d 1360, 1373 (Fed. Cir. 2010) (noting “wide latitude” in admitting expert testimony, affirming court’s ruling where there was an “adequate relationship” between expert’s experience and issues).

Next, petitioners disagree with the substance of Dr. Raymond’s conclusions, citing three reasons why there is insufficient evidence of the proposed relationship between Amelia’s SCN1A mutation and her SMEI.

<sup>3</sup> Once the petitioner has established a prima facie case of causation under the Vaccine Act, the burden shifts to the government to “show by a preponderance of the evidence that the injury is due to [a] factor[ ] unrelated to the vaccine.” *Broekelschen*, 618 F.3d at 1342 (citing 42 U.S.C. § 300aa-13(a)(1)(B)). It is not enough for a factor unrelated to be a “substantial cause” of the petitioner’s injury; rather, the factor unrelated must be “the *sole* substantial factor in bringing about the injury.” *DeBazan v. Sec’y of Health & Human Servs.*, 539 F.3d 1347, 1354 (Fed. Cir. 2008) (emphasis added); see 42 U.S.C. § 300aa-13(a)(2)(B) (requiring that factor unrelated be “principally responsible for causing the petitioner’s illness”). In determining whether a factor unrelated has been proven, the Court must “ascertain[ ] whether a sequence of cause and effect is ‘logical’ and legally probable, not [whether it is] medically or scientifically certain.” *Hanlon v. Sec’y of Health and Human Servs.*, 191 F.3d 1344, 1348 (Fed. Cir. 1999) (quoting *Knudsen v. Sec’y of Health & Human Servs.*, 35 F.3d 543, 548-49 (Fed. Cir. 1994)).

First, petitioners complain that “the SCN1A variant that Amelia Stone has is **both** novel and unstudied.” (Mot. at 29 (emphasis in original).) Dr. Raymond “agreed [with petitioners] ... that evidence of Amelia’s specific genotype-phenotype<sup>4</sup> does not currently exist.” (Dec. at 33.) However, Dr. Raymond explained that “similar mutations in nearby areas [of the relevant gene] are documented,” and “[t]he phenotype for these reported mutations is predominantly SMEI.” (*Id.*) Moreover, Dr. Raymond was able to infer the genotype-phenotype relationship from a number of factors that, “when examined by a geneticist, *cumulatively* demonstrate that Amelia’s SMEI is caused by her SCN1A mutation.” (*Id.* at 26 (emphasis in original).) Dr. Raymond’s analysis of those factors was well-supported by medical literature, as detailed in the special master’s decision. (*See id.* at 22-26, 31-35.)

Second, petitioners argue that it is impossible to tell whether the SCN1A mutation caused Amelia’s SMEI because studies show that “even people who have the **exact** same SCN1A variant have very different clinical outcomes.” (Mot. at 30 (emphasis in original).) However, the studies cited by petitioners involve people with SCN1A mutations that are significantly different from and therefore “not comparable” to Amelia’s SCN1A mutation. (*See* Dec. at 37-38.) In any event, the special master found that “neither petitioners’ counsel nor Dr. Kinsbourne is qualified to analyze this literature and discuss its significance in relation to the facts of this case.” (*Id.* at 38.) Indeed, the special master found that “Dr. Kinsbourne has no expertise in the field of genetics,” which “significantly limited his ability to offer reliable, persuasive, and cogent testimony in this case.” (*Id.* at 12.) Although the special master gave petitioners two opportunities to offer the testimony of a geneticist (first at trial and again on remand), petitioners declined to do so, leaving Dr. Raymond’s testimony essentially “unrebutted.” (*See id.* at 10.)

Third, petitioners criticize Dr. Raymond’s use of “circular logic,” claiming that he “started with the proposition that [Amelia’s] ultimate condition was only caused by her SCN1A variant,” and therefore failed to consider whether the DTaP vaccine may also have been a cause. (Mot. at 34.) This is not an accurate characterization of Dr. Raymond’s methodology. Instead, Dr. Raymond “work[ed] backwards, starting with the change in function” and looked for “certain factors that he or any geneticist examines to determine if the mutation found in a patient is the cause of the patient’s disease.” (Dec. at 24.) Having found those factors in Amelia’s case, Dr. Raymond determined that the mutation of Amelia’s SCN1A gene was the cause of her SMEI. (*See id.* at 22-26, 31-35.) Moreover, Dr. Raymond considered but rejected the possibility that the DTaP vaccine may have been a cause, citing studies where mice with SCN1A mutations developed SMEI despite never having been exposed to the DTaP vaccine. (*See id.* at 47.) Based on the foregoing, the Court finds that the special master did not act arbitrarily or capriciously in accepting Dr. Raymond’s testimony that an SCN1A mutation was the sole cause of Amelia’s SMEI.

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<sup>4</sup> A phenotype is an “observable expression of genes,” such as eye or hair color. LEE L. RUSS, ET AL., ATTORNEYS MEDICAL ADVISOR § 13:2 (2010). A genotype is a “person’s actual genetic makeup with regard to any given trait.” *Id.* “Genotypes are the particular combinations of genes which manifest as phenotypes.” *Id.*

## B. DTaP Vaccine as a Substantial Cause of Amelia's SMEI

Petitioners argue that even if there was a genetic basis for Amelia's SMEI, "[t]he record in this case does not permit a **legal conclusion** that Amelia Stone's DTaP vaccination was not a cause of her SMEI." (Mot. at 19 (emphasis in original).) Contrary to petitioners' claim, the record in this case does permit such a conclusion.<sup>5</sup> Dr. Kinsbourne claimed that although SMEI has a "genetic bas[is]," the vaccine-induced fever worsened Amelia's SMEI by lowering her seizure "threshold." (Dec. at 26.) Petitioners claim that lowering the seizure threshold means the propensity to seize has increased, and the child is more likely to have seizures in the future. (Trans. from Hearing on Mot. for Review at 10, Sept. 23, 2010.) However, the special master found that the medical literature relied upon by Dr. Kinsbourne did not support his claim. (*See* Dec. at 37-41.) Instead, the special master found that the articles relied upon by Dr. Kinsbourne "either do not ultimately stand for the proposition he stated, are dated, include studies of limited application, or [...] discuss [a different vaccine]." (*See id.* at 46.) Indeed, one of the articles cited by Dr. Kinsbourne directly contradicts his testimony, concluding that "the role of vaccination as a significant trigger for encephalopathy is *unlikely* for several reasons." (*Id.* at 44 (quoting S.F. Berkovic, *et al.*, *De novo mutations of the sodium channel gene SCN1A in alleged vaccine encephalopathy: a retrospective study*, 5 *Lancet Neurology* 488, 491 (2006)) (emphasis added).) The special master was therefore not arbitrary or capricious in finding that petitioners failed to prove the DTaP vaccination was a substantial cause of Amelia's SMEI.<sup>6</sup>

## C. Initial Vaccine-Induced Seizures as a Cause of Brain Damage

Although the parties disagree as to whether the DTaP vaccination affected Amelia's SMEI, they agree that the vaccination caused a fever which, in turn, caused Amelia's first two seizure episodes. Petitioners argue that Amelia suffered brain damage as a result of her two initial seizure episodes, thus entitling her to compensation under the Vaccine Act. In support of their argument, petitioners cite two medical records from Amelia's August 28, 2001 visit to the hospital. (*See* Ex. 2 at 7 (noting "almost no movement in her left upper extremity" and "left-sided floppiness"); Ex. 7(a) at 103 (noting that EEG<sup>7</sup> was "abnormal due to background

<sup>5</sup> In order to prevail in a non-table case, petitioners must prove by a "preponderance of the evidence" that the vaccine was "not only a but-for cause of the injury but also a substantial factor in bringing about the injury." *Cedillo*, 617 F.3d at 1338 (quoting *Shyface v. Sec'y of Health & Human Servs.*, 165 F.3d 1344, 1352 (Fed. Cir. 1999)).

<sup>6</sup> Petitioners claim that Dr. Raymond and Dr. Kohrman (another expert witness for respondent) admitted that Amelia's vaccine "caused the onset of her seizures." (Mot. at 19-20.) Petitioners are mistaken. Dr. Raymond expressly denied that the vaccine caused Amelia to "go[ ] on to have Dravet syndrome." (*Id.* (quoting Tr. R. 2 at 334).) Dr. Kohrman admitted only that the vaccine was "likely what caused the fever that triggered the initial seizure," not that it caused or worsened Amelia's SMEI. (*Id.* (quoting Tr. R. at 40).)

<sup>7</sup> EEG stands for electroencephalography, which is "a method of amplifying, recording and analyzing the electrical impulses of the brain." RUSS, ET AL., ATTORNEYS MEDICAL ADVISOR § 20:17.

asymmetry with left-sided slowing,” suggesting “neurophysiologic dysfunction.”).) However, neither of these records indicate that Amelia suffered any brain damage as a result of her initial seizures. To the contrary, Amelia’s medical records from August 31, 2001 state that Amelia “continued to feed well and showed no seizure activity or neurologic deficit” (Ex. 7(a) at 107), and that she had a “[n]ormal head MRI for age with no findings to explain patient’s symptoms” (*id.* at 94). Indeed, petitioners’ own expert failed to testify that Amelia actually suffered brain damage from her initial seizures, stating only that the seizures were “potentially damaging” and conceding that “[a] trigger doesn’t necessarily have to have a further deeper impact.” (Tr. R. 2 at 482.) Thus, the special master’s determination that “there is no evidence the initial seizure[s] injured the brain” (Dec. at 52) was neither arbitrary nor capricious.<sup>8</sup>

#### **D. The Special Master’s Application of Legal Standards**

Petitioners argue that the special master misapplied a number of legal standards relating to superseding cause, petitioners’ burden of proof, and section 11(c) of the Vaccine Act. None of these arguments are availing to petitioners.

First, petitioners argue that Amelia Stone’s SCN1A gene mutation was not a superseding cause of Amelia’s SMEI.<sup>9</sup> Petitioners’ argument misapprehends the nature of the special master’s decision. The special master did not find that the SCN1A gene mutation was a superseding cause of Amelia’s SMEI; indeed, the word “superseding” never appears in the decision. Rather, the special master found that the SCN1A gene mutation was the sole cause and was principally responsible for Amelia’s SMEI, and that the vaccine had no causal affect whatsoever. (*See* Dec. on Remand at 2-4.)

Next, petitioners argue that the special master erred by requiring petitioners to demonstrate how Amelia’s vaccine injury damaged her brain. Petitioners are correct that “a Vaccine Act claimant is not required to present proof of causation to the level of scientific certainty.” *Moberly v. Sec’y of Health & Human Servs.*, 592 F.3d 1315, 1324 (Fed. Cir. 2010). However, “the special master is entitled to require some indicia of reliability to support the assertion of the expert witness;” the claimant must provide at least some evidence “that the proposed mechanism was at work” in the case before the Court. *Id.* As set forth in sections III.A. and B., petitioners failed to adduce evidence in support of Dr. Kinsbourne’s assertions.

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<sup>8</sup> Petitioners claim that 42 U.S.C. § 300-aa(11)(c)(i)(D) entitles them to compensation because Amelia was hospitalized as a result of her two initial seizures. (*See* Mot. at 35.) However, petitioners have failed to cite any request for such compensation in the proceedings before the special master. Because “[a]ny fact or argument not raised specifically in the record before the special master will be considered waived and cannot be raised by either party in proceedings on review of a special master’s decision,” the Court will not consider this argument. RCFC Appx. B, Vaccine Rule 8(f)(1).

<sup>9</sup> A superseding cause “is an act of a third person or other force which by its intervention prevents the actor from being liable for harm to another which his antecedent negligence is a substantial factor in bringing about.” RESTATEMENT (SECOND) OF TORTS, §440 (2010).

Finally, petitioners argue that they are entitled to compensation because “[e]very provision of subsection 11(c) of the Vaccine Act<sup>10</sup> has been satisfied and, in fact, conceded.” (Mot. at 35.) However, section 13 of the Vaccine Act allows the government to escape liability if the complained-of injury “is due to factors unrelated to the administration of the vaccine.” 42 U.S.C. §300aa-13(a)(1)(B). Although petitioners were initially able to make a prima facie case for compensation under section 11, the special master ultimately determined that Amelia’s SMEI was caused solely by an SCN1A gene mutation, a factor unrelated to the vaccine. (*See* Dec. on Remand at 2; Dec. at 11.) The special master therefore applied the correct legal standards in his decision on remand.

The Court has considered petitioners’ various other arguments, and finds them to be without merit.

#### IV. CONCLUSION

For the reasons set forth above, petitioners’ motion for review is denied, and the special master’s opinion on remand is affirmed. The Clerk of the Court is hereby directed to enter judgment accordingly.

s/ Lawrence S. Margolis  
LAWRENCE S. MARGOLIS  
Senior Judge, U.S. Court of Federal Claims

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<sup>10</sup> Section 11(c) sets out the elements that a petitioner must prove in order to obtain compensation under the Vaccine Act. *See* 42 U.S.C. § 300aa-11(c).